



RUNX2 gene

runt related transcription factor 2

Normal Function

The *RUNX2* gene provides instructions for making a protein that is involved in bone and cartilage development and maintenance. This protein is a transcription factor, which means it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Researchers believe that the *RUNX2* protein acts as a "master switch," regulating a number of other genes involved in the development of cells that build bones (osteoblasts).

Health Conditions Related to Genetic Changes

cleidocranial dysplasia

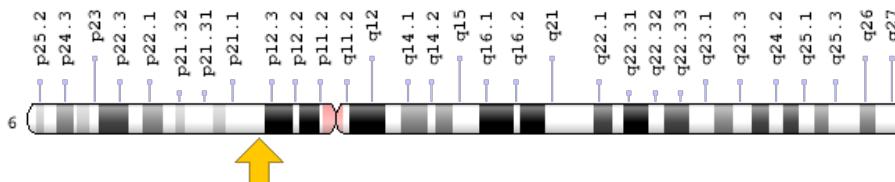
More than 70 mutations in the *RUNX2* gene have been identified in individuals with cleidocranial dysplasia. Some mutations change one protein building block (amino acid) in the *RUNX2* protein. Other mutations introduce a premature stop signal that results in an abnormally short protein. Occasionally, the entire gene is missing.

These genetic changes reduce or eliminate the activity of the protein produced from one copy of the *RUNX2* gene in each cell, decreasing the total amount of functional *RUNX2* protein. This shortage of functional *RUNX2* protein interferes with normal bone and cartilage development, resulting in the signs and symptoms of cleidocranial dysplasia. In rare cases, affected individuals may experience additional, unusual symptoms resulting from the loss of other genes near *RUNX2*.

Chromosomal Location

Cytogenetic Location: 6p21.1, which is the short (p) arm of chromosome 6 at position 21.1

Molecular Location: base pairs 45,328,142 to 45,664,032 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- acute myeloid leukemia 3 protein
- AML3
- CBF-alpha 1
- CBFA1
- CCD
- CCD1
- core-binding factor, runt domain, alpha subunit 1
- MGC120022
- MGC120023
- OSF2
- osteoblast-specific transcription factor 2
- PEA2aA
- PEBP2A1
- PEBP2A2
- PEBP2aA
- PEBP2aA1
- polyomavirus enhancer binding protein 2 alpha A subunit
- runt-related transcription factor 2

- RUNX2_HUMAN
- SL3-3 enhancer factor 1 alpha A subunit
- SL3/AKV core-binding factor alpha A subunit

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Osteogenesis: The Development of Bones
<https://www.ncbi.nlm.nih.gov/books/NBK10056/>

GeneReviews

- Cleidocranial Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK1513>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RUNX2%5BTIAB%5D%29+OR+%28runt-related+transcription+factor+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

OMIM

- RUNT-RELATED TRANSCRIPTION FACTOR 2
<http://omim.org/entry/600211>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/RUNX2ID42183ch6p21.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RUNX2%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10472
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/860>
- UniProt
<http://www.uniprot.org/uniprot/Q13950>

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